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Brief Clinical Report

Cerebrofaciothoracic Syndrome

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We report on a patient with a large septum pellucidum, hypodensity of gray matter, hypertelorism, and costovertebral anomalies. Only 5 previous cases have been described with this distinctive phenotype. Autosomal recessive inheritance seems likely. © 1996 Wiley-Liss, Inc.

KEY WORDS: hypertelorism, costovertebral anomalies, CNS anomalies

INTRODUCTION

We have recently seen a patient whose manifestations are compatible with cerebrofaciothoracic syndrome. Only a few cases have been previously recorded. A summary of our patient and a table comparing him with previously reported cases (Table I) appear below.

CLINICAL REPORT

This male infant with multiple congenital anomalies was born at term. Birth weight was 4,160 g (90th centile), birth length 53 cm (90th centile), and OFC 36 cm (75th centile). The pregnancy was normal. Mother and father were normal and consanguinity was denied. Karyotype was normal.

Examination at 5 weeks showed striking anomalies (Figs. 1–4). He had a wide anterior fontanelle, frontal upsweep with poliosis, forehead wrinkling, hypertelorism, upslanting palpebral fissures, sparse eyebrows and eyelashes, short broad nose, unilateral cleft lip-palate, micrognathia, and apparently low-set posteriorly angulated ears, short neck, narrow shoulders, atrial septal defect, supernumerary nipple, inguinal hernia, and postaxial polydactyly of the hands. He was hypotonic with a severe swallowing deficit. Radiographs showed costovertebral anomalies. CT scan showed large septum pellucidum and diffuse hypodensity of gray matter. Forehead wrinkling disappeared by 8 months but synophrys, not evident at birth, was present.

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COMMENT

The distinct phenotype in our patient was reported previously by Pascual-Castroviejo et al. [1975] and

TABLE I. Manifestations in Cerebrofaciothoracic Syndrome

Manifestation	Previously reported cases	Our patient
Growth		
High birth weight (>90th centile)	1/2	+
Postnatal growth deficiency	1/1	–
Performance		
Mental deficiency	4/4	[a]
Affable behavior	4/4	?
Central nervous system		
Hypoplasia of corpus callosum	2/4	–
Large septum pellucidum	2/4	+
Cyst of septum pellucidum	1/4	–
Calcification of petroclinoid ligament	3/5	–
Diffuse hypodensity of gray matter	0/5	+
Craniofacial		
Macrocephaly	3/5	–
Brachycephaly	3/5	–
Hypertelorism	5/5	+
Epicanthic folds	3/5	–
Synophrys	3/5	+
Broad short nose	5/5	+
Cleft lip-palate	1/5	+
Micrognathia	1/2	+
Low-set, posteriorly angulated ears	5/5	+
Short neck	5/5	+
Skeletal		
Costovertebral anomalies	5/5	+
Narrow thorax/shoulders	3/5	+
Raised scapulae	3/5	–
Postaxial manual polydactyly	0/5	+
Other		
Poliosis	0/5	+
Inguinal hernia	2/5	+
Sacral dimple	1/5	–
Supernumerary nipple	0/5	+
Shawl scrotum	1/5	–
Atrial septal defect	0/5	+

[a]Hypotonia and severe swallowing deficit.

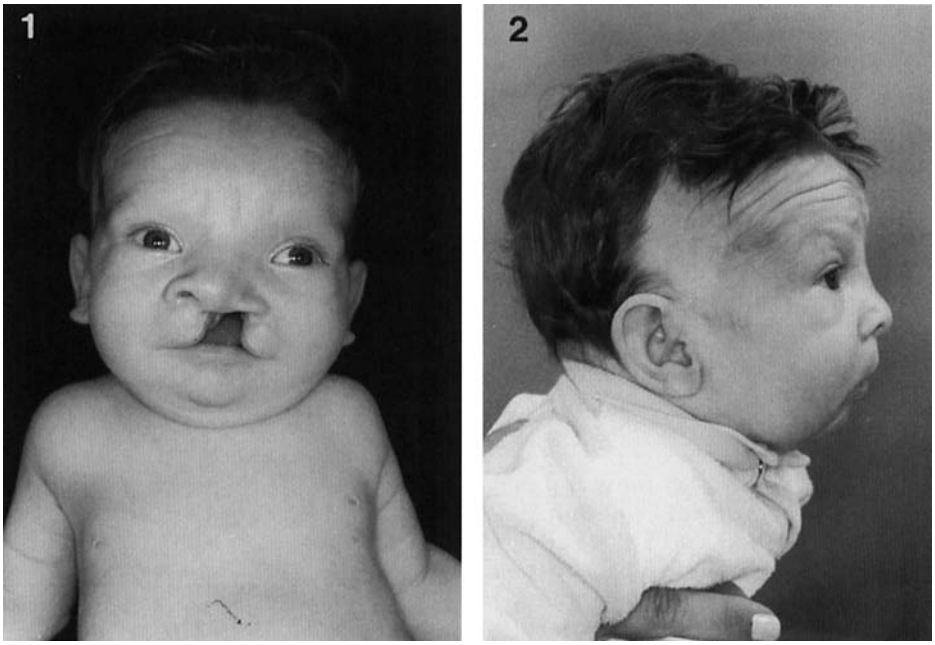

Fig. 1. Hypertelorism, sparse eyebrows, upslanting palpebral fissures, short broad nose, cleft lip-palate.
Fig. 2. Forehead wrinkling, sparse eyebrows, apparently low-set, posteriorly angulated ears, micrognathia.

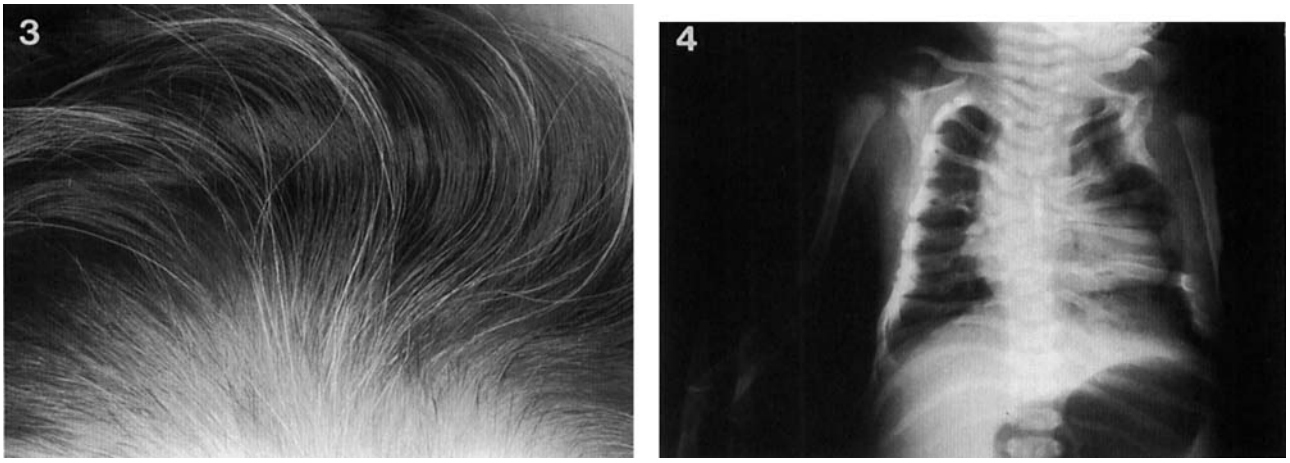

Fig. 3. Frontal upsweep with poliosis.
Fig. 4. Costovertebral anomalies.

Philip et al. [1992], and 5 cases have been recorded to date. Two affected sibs[1] of a consanguineous union were noted by Philip et al. [1992]. Thus, autosomal recessive inheritance seems likely.

Cerebrofaciothoracic syndrome can be distinguished from autosomal recessive spondylocostal dysostosis [Casamassima et al., 1981], autosomal recessive Robinow syndrome [Gorlin et al., 1990], and the lethal chondrodysplasia with spondylocostal dysostosis described by Moerman et al. [1985].

[1] Both pregnancies were complicated by hydramnios.

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